TREM2 Gene

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Triggering receptor expressed on myeloid cells 2: The TREM2 gene provides instructions for making a protein called triggering receptor expressed on myeloid cells 2.

genes

1. Normal Function

The *TREM2* gene provides instructions for making a protein called triggering receptor expressed on myeloid cells 2. As its name suggests, this protein is made in myeloid cells, which are cells produced in bone marrow. The TREM2 protein is found on the cell surface, where it interacts with the protein produced from the *TYROBP* gene. The TREM2 and TYROBP proteins form a complex that transmits chemical signals to activate the cell.

The TYROBP-TREM2 complex was first identified in the immune system. This complex is involved in the growth and development of several types of immune cells, particularly dendritic cells. The TYROBP-TREM2 complex activates these cells, triggering an inflammatory response to injury or disease.

The TYROBP-TREM2 complex also activates cells in the skeletal system and in the brain and spinal cord (central nervous system). In the skeletal system, the complex is found in osteoclasts, which are specialized cells that break down and remove (resorb) bone tissue that is no longer needed. These cells are involved in bone remodeling, which is a normal process that replaces old bone tissue with new bone. In the central nervous system, the complex appears to play an important role in immune cells called microglia. These cells protect the brain and spinal cord from foreign invaders and remove dead nerve cells and other debris. Although the TYROBP-TREM2 complex plays a critical role in osteoclasts and microglia, its exact function in these cells is unclear

2. Health Conditions Related to Genetic Changes

2.1. Polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy

At least 10 mutations in the *TREM2* gene have been identified in people with polycystic lipomembranous osteodysplasia with sclerosing leukoencephalopathy (commonly known as PLOSL). Some mutations prevent the cell from making any TREM2 protein, while others result in the production of an abnormally short, nonfunctional

version of the protein. Still other mutations change the structure of the TREM2 protein, preventing it from reaching the cell surface.

Researchers believe that the signs and symptoms of PLOSL are related to defective TYROBP-TREM2 signaling in osteoclasts and microglia. The bone abnormalities seen with this disorder are probably related to malfunctioning osteoclasts, which are less able to resorb bone tissue during bone remodeling. In the central nervous system, defective signaling through the TYROBP-TREM2 complex causes widespread abnormalities of microglia. Researchers are working to determine how these abnormalities lead to the neurological problems associated with PLOSL.

3. Other Names for This Gene

- TREM-2
- TREM2_HUMAN
- Trem2a
- Trem2b
- Trem2c
- triggering receptor expressed on monocytes 2
- triggering receptor expressed on myeloid cells 2a

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